Testimony in Support of HB6580 An Act Creating an Advisory Council on Rare Diseases

I was in a sleep deprived haze, similar to all first time mothers who bring their newborns home, when I received the call from my daughter's pediatrician's office stating something popped up on her newborn screening test and a genetics' counselor would be following up with me within the next few days.

I hung up the phone puzzled as to why a counselor would be calling me about my 8-day-old perfect little pink bundle of joy. Did they just want to make sure she was happy and nursing? Maybe she seemed fussy in the hospital? Why else would a newborn need a counselor?

What didn't click was the genetic's part; it was quickly explained to me as the phone rang 15 minutes later that the counselor on the phone was not a psychiatric one but one that deals with inborn errors of metabolism and one of my daughter's newborn screening markers, specifically, her long chain fats, were slightly off. I was assured that it was probably a false positive as the Ohio cutoff was .09 units of measure and hers was .0913.

After a blood draw and expedited testing over the next 24 hours we found out we had won the rare disease genetics lottery- our daughter has CPT2, a long chain fatty acid disorder that didn't allow her body to convert fat into energy properly and solely relies on glucose for energy.

Yeah, I didn't know what that meant either. Especially while in still in my sleep-deprived haze. I was told to never let her fast for more than 2-3 hours until she hit the 3-month mark; this wasn't a real issue since I was EBF. Until the 3-month mark, we had bi-weekly check-ups to see how she was progressing.

We were one of the lucky ones, our daughter inherited the S113L mutation on her first chromosome, the "mild" version of the disease, which could be easily controlled by diet and avoiding fasting.

How hard could that be correct?

Until your brain quickly jumps to the question of "how is she supposed to sleep through the night ever?"

"What if she gets a stomach bug and keeps throwing up?"

"What if she's a picky eater and refuses to eat just to assert the normal control a 3 year old does?"

Five years later, I'm still learning "hard" was an understatement.

The easy part was limiting her diet to 15 grams of fat/1000 calories/day; the easy part was at 13 months mixing raw cornstarch into her applesauce to help maintain her glucose up to 12 hours allowing her to get a full night's sleep right before bedtime; the easy part was substituting medium chain fats via a special oil (which I want to mention costs about $100/bottle and is not covered by insurance; a bottle lasts about a month) into her food vs. the long chain fats her body couldn't metabolize (such as canola oil, vegetable oil, olive oil).

The hard part was keeping her healthy; I was fortunate as she did not get her first cold until 10 months because of my continued nursing efforts and being with my mother during the day vs. a daycare.

At 18 months, however, she wasn't gaining the weight the doctors wanted her to, as she just wasn't that "into" food; she always preferred breast milk and that wasn't an option for her at that age. I was told it was fairly common and some kids were just not "eaters"; however while normal children could be left to continue their "rabbit grazings" as her doctor called it, my daughter couldn't. It could be catastrophic for her. Their recommendation was to put a feeding tube into her stomach and feed her continuously through that.

My reaction was simple "over my dead body; I'll chase her with a banana if I have to for the next three years". And that's what I did; I quit a very lucrative career in finance and literally chased her around with a banana (and other non fatty foods) for the next year until I got her into the 50th percentile of weight.

I was a proud mama; I did my job and I did it well.

Then preschool started; or in other words, the illnesses started. ONE AFTER ANOTHER AFTER ANOTHER.

So what you're thinking right? Kids always get sick, it's normal. But remember the fasting thing from before? How many of us want to eat when we have a 102 fever? All we want to do is sleep and rest. Except children like my daughter cannot do that; they will fall into a hypoglycemic coma and go into organ failure. Either you get them to drink sugary drinks or bring them to a hospital for an IV of dextrose.

We did the latter once; I'm sure she'll never remember the traumatic 2 day hospital stay thank G-d. And I will never forget it.

So every time she gets sick, her sugar and calorie consumption have to be maintained. At times of severe illness, such as fever viruses, pneumonia, croup, bronchitis, I pipet ginger ale into her cheek every two hours while she's sleeping with a syringe to avoid waking her yet giving her the necessary sugars to maintain her glucose through the night to avoid a metabolic crisis and hence, a hospital stay.

The act itself is exhausting. The anxiety and fear that accompanies it is indescribable. As she has gotten older, it has become easier. Or maybe we just have become used to it.

I still haven't returned to the working world since every time a stomach bug or the hand-foot-mouth virus goes around the classroom, I keep her home to minimize her exposure to it. This translates into about a month or more, of missed school each year. And that's not counting the times that she herself is actually sick.

And our lives are filled with a constant "what if's?"

What if we take that Disney Cruise and a GI virus breaks out?

What if her classmate is infected with the hand-foot-mouth virus his little sister just had and isn't exhibiting symptoms yet and sits next to my daughter at lunchtime?

What if the table we were just seated for dinner at wasn't properly cleaned and the live flu is still lingering around right where my daughter is to be seated?

We wouldn't have to be always asking those what if's if magically, her "S" and "L" amino acids were switched as they should be on marker 113 on chromosome 1.

The answer is SO easy; how to get there is not.

It is estimated that between 300-800 people in the WORLD have what my daughter has.

There is no funding for it; there is barely any medical knowledge about it; pediatrician's and emergency doctors are CLUELESS about it because essentially NOTHING is taught about it during medical school or residency training. They get more education I've been told by my doctor friends on male erectile issues than these types of metabolic disorders.

Perhaps this hearing and committee can help me stop asking my what-if's one day.